



IDS gene

iduronate 2-sulfatase

Normal Function

The *IDS* gene provides instructions for producing an enzyme called iduronate 2-sulfatase (I2S), which is essential for the breakdown of large sugar molecules called glycosaminoglycans (GAGs). Specifically, I2S removes a chemical group known as a sulfate from a molecule called sulfated alpha-L-iduronic acid, which is present in two GAGs called heparan sulfate and dermatan sulfate. I2S is located in lysosomes, compartments within cells that digest and recycle different types of molecules.

Health Conditions Related to Genetic Changes

mucopolysaccharidosis type II

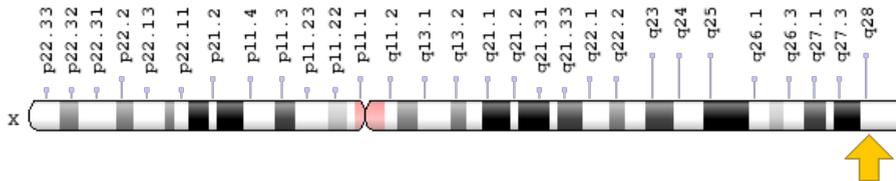
More than 300 mutations in the *IDS* gene have been found to cause mucopolysaccharidosis type II (MPS II). Mutations that change one DNA building block (nucleotide) are the most common. All mutations that cause MPS II reduce or completely eliminate the function of I2S. It usually cannot be determined whether a certain mutation will cause severe or mild MPS II; however, mutations that result in the complete absence of I2S cause the more severe form of the disorder.

Lack of I2S enzyme activity leads to the accumulation of heparan sulfate and dermatan sulfate within cells, specifically inside the lysosomes. The buildup of these GAGs increases the size of the lysosomes, which is why many tissues and organs are enlarged in MPS II. Researchers believe that the accumulated GAGs may also interfere with the functions of other proteins inside the lysosomes and disrupt the movement of molecules inside the cell.

Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 149,476,990 to 149,505,354 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- IDS_HUMAN
- iduronate-2-sulfatase
- iduronate 2-sulfatase (Hunter syndrome)

Additional Information & Resources

Educational Resources

- Eureka Bioscience Collection: Defects in Glycosaminoglycan Degradation (Mucopolysaccharidoses)
<https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53462>

GeneReviews

- Mucopolysaccharidosis Type II
<https://www.ncbi.nlm.nih.gov/books/NBK1274>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28iduronate+2-sulfatase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- MUCOPOLYSACCHARIDOSIS, TYPE II
<http://omim.org/entry/309900>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_IDS.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=IDS%5Bgene%5D>
- HGNC Gene Family: Sulfatases
<http://www.genenames.org/cgi-bin/genefamilies/set/410>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5389
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3423>
- UniProt
<http://www.uniprot.org/uniprot/P22304>

Sources for This Summary

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Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/IDS>

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